Quantitative Genetics - Multiple Loci

Peter von Rohr

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Two and more Loci

- Two loci A and B having an effect on the same quantitative trait.
- Additional effect in genetic model: **Interaction** effect (also known as epistasis)
- Interaction occurs if effect of one locus can have an influence on the effect of the other locus
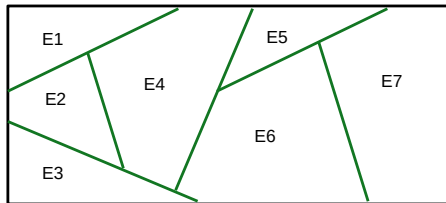- Interaction is quantified by

$$I_{AB} = V - V_A - V_B$$

Influence of Two Loci on Quantitative Trait

Two Loci Influencing a Quantitative Trait

Locus A

Locus B

I_{AB}

$V_A = \mu_A + BV_A + D_A$

$V_B = \mu_B + BV_B + D_B$

$V = V_A + V_B + I_{AB}$
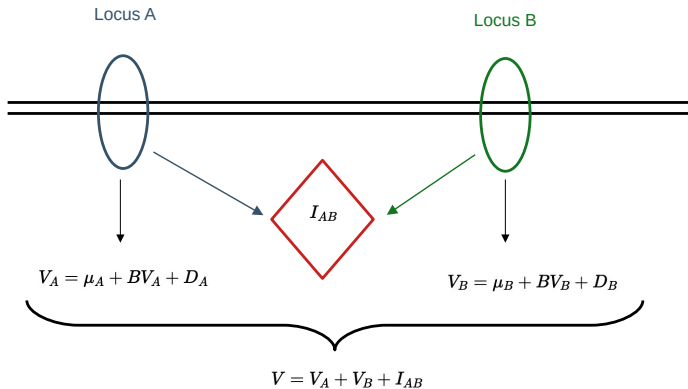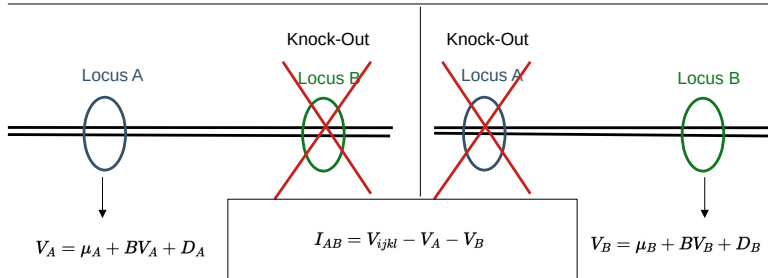
Quantify Interaction Effect

For Animal d with genotype:
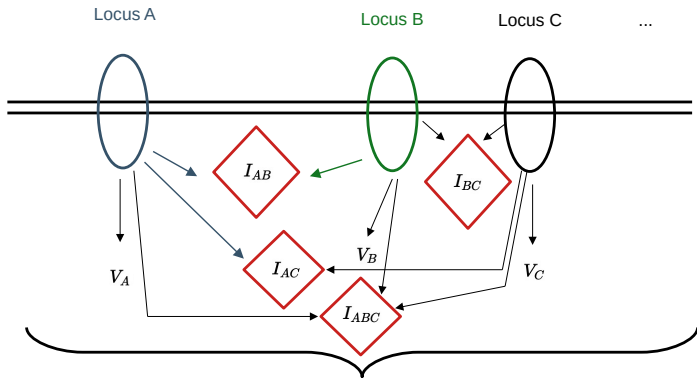$A_i A_j B_k B_l$

Total Genetic Value V_{ijkl}
as mean phenotypic value (Y)
accross different environments
(E1 to E7):

$$V_{ijkl} = \frac{1}{N} \sum_{m=1}^{N} Y_{ijklm}$$

Knock-Out

Locus A

Knock-Out

Locus B

Locus A

Locus B

$V_A = \mu_A + BV_A + D_A$

$$I_{AB} = V_{ijkl} - V_A - V_B$$

$V_B = \mu_B + BV_B + D_B$

Many Loci Influencing a Quantitative Trait

$$V = V_A + V_B + V_C + \ldots + I_{A \cdot B \cdot C \cdot \ldots}$$

with $\quad I_{A \cdot B \cdot C} = I_{AB} + I_{AC} + I_{BC} + I_{ABC}$

Summary Genetic Model

- Given genetic model

$$V = V_A + V_B + V_C + ... + I_{A \cdot B \cdot C \cdot ...}$$

- Decomposition

$$V = \mu_A + BV_A + D_A + \mu_B + BV_B + D_B + \mu_C + BV_C + D_C + ... + I_{A \cdot B \cdot C \cdot ...}$$

- Re-group

$$V = \mu_A + \mu_B + \mu_C + ... + BV_A + BV_B + BV_C + ...$$
$$+ D_A + D_B + D_C + ... + I_{A \cdot B \cdot C \cdot ...}$$

- Collect

$$V = \mu + BV + D + I_{A \cdot B \cdot C \cdot ...}$$

Extend to Phenotypes

▶ We know from earlier

$$Y = V + E$$

▶ Insert decomposition of V

$$Y = \mu + BV + D + I_{A \cdot B \cdot C \cdots} + E$$

▶ For breeding, only BV is important

$$Y = \mu + BV + E^*$$

with $E^* = D + I_{A \cdot B \cdot C \cdots} + E$

Split of Environment

Non-genetic environment is split into

- ▶ known part, corresponding to additional information on animal, like herd, age, sex, . . . leads to vector of fixed effects and regression coefficients $\rightarrow b$
- ▶ unknown part leads to random resiudal part $\rightarrow e$

Model of Phenotypic Observation

Phenotypic observation (y_i) of an animal i can be modelled as

$$y_i = x_i^T \cdot b + u_i + e_i$$

where

y_i	phenotypic observation of animal i
x_i	vector of regression covariates or indicator values for fixed effects
b	vector of unknown fixed effects and unknown regression coefficients
u_i	random breeding value of animal i
e_i	random environmental effects of animal i

Aggregation over Population

Take all observations from all animals in a given population

$$y = X \cdot b + Z \cdot u + e$$

where

y	vector of phenotypic observations of all animals
b	vector of unknown fixed effects and unknown regression coefficients
u	vector of random breeding values of all animals
e	vector of random environmental effects
X	design matrix with regression covariates or indicator values for fixed effects
Z	design matrix relating breeding values to observations

Example Dataset I

Animal	Sire	Dam	Herd	Weaning Weight	Breast Circumference
14	1	5	1	2.44	1.48
15	1	5	1	2.41	1.47
16	1	6	2	2.51	1.50
17	1	6	2	2.55	1.47
20	2	8	1	2.34	1.46
21	2	8	1	1.99	1.38

Example Dataset II

- ▶ Weaning weight as response variable y
- ▶ Breast circumference as regression variable
- ▶ Herd as fixed effect
- ▶ Breeding values for
  - ▶ animals with observations: own-performance
  - ▶ sires: sire model
  - ▶ all animals in pedigree: animal model